Opis choroby *

Definicja

A rare subtype of autosomal dominant limb-girdle muscular dystrophy ,with a variable age of onset, characterized by progressive, proximal weakness and wasting of the shoulder and pelvic musculature (with the pelvic girdle, and especially the ileopsoas muscle, being more affected) and frequent association of calf hypertrophy, dysphagia, arachnodactyly with or without finger contractures and/or distal and axial muscle involvement. Additional features include an abnormal gait, exercise intolerance, myalgia, fatigue and respiratory insufficiency. Cardiac conduction defects are typically not observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant limb-girdle muscular dystrophy type 1F
LGMD1F
LGMD type 1F
LGMD1F
Limb-girdle muscular dystrophy type 1F

Kod ORPHA
55595

Kod OMIM
608423

Kod ICD10
G71.0

Kod ICD11
8C70.40

---

*Źródło

orphanet